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Original Research

Prenatal Diagnosis of Fetal Visceral Situs Abnormalities: A Retrospective Study

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Abstract

Background: To explore the necessity for prenatal diagnosis and evaluate related diagnostic methods for fetal visceral situs abnormalities. **Methods**: A retrospective analysis was performed to examine the clinical data and pregnancy outcomes of 43 cases of fetal visceral situs abnormalities diagnosed at our hospital between May 2018 and September 2023, using prenatal ultrasound consultation as the diagnostic standard. **Results**: Among the 43 cases of fetal visceral situs abnormalities, 46.51% (20/43) were diagnosed as situs inversus, of which 95.00% (19/20) had no associated cardiac structural abnormalities. Heterotaxy syndrome accounted for 53.49% (23/43) of cases and was associated with cardiac structural abnormalities (100.00%, 23/23) of these patients. The most common cardiac abnormalities involved a single atrium and a single ventricle (56.52%, 13/23), followed by double-outlet right ventricle (34.78%, 8/23), and pulmonary artery stenosis (30.43%, 7/23). Amniocentesis was performed in 21 (48.84%, 21/43) cases, with both chromosomal karyotyping and chromosomal microarray analyses yielding negative results. Among these cases, 15 underwent whole-exome sequencing (WES), which identified 5 with suspected pathogenic gene variants related to primary ciliary dyskinesia (PCD) and 1 with Holt–Oram syndrome (HOS). **Conclusions**: Prenatal ultrasonography is a reliable method for diagnosing fetal visceral situs abnormalities. Moreover, for fetuses diagnosed with visceral situs abnormalities and pregnant women with normal chromosomal results, WES remains essential to improve the detection rates of PCD or other genetic abnormalities.

Keywords: abnormal position of viscera; WES; prenatal diagnosis

1. Introduction

Fetal visceral situs abnormalities are rare congenital anomalies, occurring in approximately 1/2500 to 1/20,000 in the population. These anomalies are broadly classified as complete situs inversus (situs inversus totalis) and partial situs inversus (heterotaxy syndrome) [1]. Partial situs inversus is even rarer and is frequently associated with complex malformations. Studies have shown the potential relationship of fetal visceral situs abnormalities with genetics, gene mutations, and chromosomal structural aberrations [2,3], which significantly affect postnatal survival rates and long-term quality of life [4,5]. This study retrospectively analyzed the prenatal clinical data of 43 pregnant women to explore the need for prenatal diagnosis and related diagnostic methods for fetal visceral situs abnormalities to provide an informed basis for prenatal management and assist patients in making prenatal decisions.

2. Methods

2.1 Study Design and Participants

This study included 43 women diagnosed with fetal visceral situs abnormalities by prenatal ultrasound examination between May 2018 and September 2023 at the Prenatal Diagnosis Consultation Center of International Peace Maternity and Child Health Hospital, Shanghai Jiao Tong University School of Medicine. These women were 19–42 (mean, 30.53 ± 5.56) years old, with gestational ages ranging from 21 to 26 weeks. Follow-up continued until pregnancy was completed, and baseline clinical data and fetal birth information were recorded.

The study was approved by the hospital's medical ethics committee and all participants provided informed consent.

2.2 Instruments and Methods

2.2.1 Instruments

High-end color Doppler ultrasound diagnostic instruments such GE Voluson E10 (GE Healthcare, Chicago, IL, USA) and Philips EPIQ IPC7 ultrasound system (Philips Healthcare, Amsterdam, Netherlands) were utilized, with probe frequencies of 2–9 and 1–5 MHz, respectively.

2.2.2 Examination Steps

(1) Determine fetal orientation based on the relationship between the fetal head and spine, identifying whether the probe's near field is on the left or right side of the fetus. (2) Continuously scan and rotate the probe to obtain a standard transverse section of the fetal upper abdomen, determining the positions of the fetal stomach bubble, inferior vena cava, abdominal aorta, and liver. Carefully observe the spleen and gastrointestinal tract. (3) Obtain a four-chamber view of the heart to determine its position in the thorax and the direction of the cardiac apex. Then, assess the atria, ventricles, and their connections to the great arteries using sequential segmental analysis while observing the morphology of the atrial appendages, continuity of the ventricular septum, and morphology, structure, and activity of the valves [6].

2.2.3 Ultrasound Diagnostic Criteria

All examinations were performed at the hospital's prenatal diagnosis consultation center by 2 experienced, certified senior ultrasound physicians.

2.3 Classification of Visceral Situs

(1) Situs solitus: normal development of the abdominal and thoracic organ positions.

(2) Situs inversus (formerly complete situs inversus): mirror image of situs solitus.

(3) Heterotaxy/isomerism syndrome (formerly partial situs inversus): left and right isomerism.

(A) Left isomerism (or polysplenia syndrome). In this type, both atria exhibit left atrial morphology (i.e., bilateral left atrial structure), often accompanied by multiple small spleens (polysplenia). The liver and stomach bubble may be in the normal (situs solitus), reversed (situs inversus), or indeterminate position. The inferior vena cava may be interrupted, with an enlarged azygos vein running posterior to the abdominal aorta. This isomerism is often associated with cardiac anomalies, such as atrioventricular septal defects or anomalous pulmonary venous drainage, as well as other organ abnormalities.

(B) Right isomerism (asplenia syndrome). In this type, both atria exhibit right atrial morphology (i.e., bilateral right atrial structure), accompanied by asplenia (absence of the spleen). The liver is typically midline in position, and the stomach bubble may be located on the left, right, or in an indeterminate position. On transverse imaging of the upper abdomen, the abdominal aorta and inferior vena cava are situated on the same side of the spine (ipsilateral arrangement). Right isomerism is often associated with complex cardiac anomalies, such as having a single ventricle or transposition of the great arteries, as well as other organ abnormalities.

2.4 Statistical Analysis

Statistical analysis was performed using IBM SPSS Statistics for Windows, version 25.0 (IBM Corp., Armonk, NY, USA). Measurement data were expressed as mean ($\bar{X}$), standard deviation (SD), and percentiles, whereas count data were presented as N (%). Fisher's exact test was employed to compare the differences in fetal visceral abnormalities accompanied by cardiac malformations. A two-sided p-value < 0.050 was considered significant.

3. Results

3.1 General Information

The study included 43 cases of fetal visceral situs abnormalities, with an estimated incidence of 0.59‰ (43/72, 499). The pregnancy outcomes included induced labor in 51.16% (22/43), normal delivery in 32.56% (14/43), and lost to follow-up in 16.28% (7/43). Natural conception accounted for 90.70% (39/43), singleton pregnancies for 97.67% (42/43), and indeterminate fetal sex for 65.12% (28/43), which was attributed to being lost to follow-up or the inability to determine the fetal sex after induced labor. Amniocentesis was performed in 48.84% (21/43) of the cases, with chromosomal karyotype and chromosomal microarray analyses yielding negative results. Among these cases, 15 underwent whole-exome sequencing (WES), and the genetic findings were as follows: 1 case carried a suspected pathogenic variant in primary ciliary dyskinesia (PCD)-related gene (maternal autosomal recessive inheritance pattern), which resulted in normal delivery; 4 had variants of uncertain significance (VUS) in PCD-related genes, which included 1 case of PCD type 3 (autosomal recessive inheritance) with pregnancy termination and 3 live births (including 2 maternal VUS and 1 *NAH9* gene variant); 1 case of Holt–Oram syndrome (HOS, autosomal dominant inheritance) with maternal VUS (likely benign), resulting in pregnancy termination; and 9 cases not showing clinically significant variants on WES (Table 1).

3.2 Abnormal Positions of Fetal Internal Organs Combined With Cardiac Structural Abnormalities

Our analysis of 43 cases of fetal visceral situs abnormalities revealed distinct patterns of cardiac anomalies between situs inversus and heterotaxy syndrome. The cohort comprised 20 (46.51%) cases of situs inversus, among which only 1 (5.00%) exhibited additional cardiac structural anomalies (pulmonary stenosis, complete transposition of great arteries, or ventricular septal defect). In contrast, all 23 cases of heterotaxy syndrome (53.49%, consistent with cardiospheric syndrome) presented with cardiovascular malformations (100.00%, 23/23), which included left (43.48%, 10/23) and right (56.52%, 13/23) isomerisms. The most prevalent anomalies were single atrium/ventricle (56.52%, 13/23), double-outlet right ventricle (34.78%, 8/23), and pulmonary stenosis (30.43%, 7/23). Statisti-

Table 1. Demographic data of 43 cases with visceral anomalies.

General information	Results
Age (years, $\bar{X} \pm$ SD)	30.53 $\pm$ 5.56
Pregnancy outcome (%/N)	
Normal delivery	32.56% (14/43)
Induced labor	51.16% (22/43)
Lost to follow-up	16.28% (7/43)
Conception method (%/N)	
Natural conception	90.70% (39/43)
IVF	9.30% (4/43)
Number of pregnancies (%/N)	
Single birth	97.67% (42/43)
Twin (DCDA)	2.33% (1/43)
Obstetric history (%/N)	
Primipara	74.42% (32/43)
Multipara	25.58% (11/43)
Fetal sex (%/N)	
Male	13.95% (6/43)
Female	20.93% (9/43)
Indeterminate	65.12% (28/43)
NIPT (%/N)	51.16% (22/43)
Chromosome negative	100.00% (22/22)
Chromosome positive	0.00% (0/22)
Amniocentesis (%/N)	48.84% (21/43)
Chromosome karyotype and icroarray analysis	
Negative	100.00% (21/21)
Positive	0.00% (0/21)

Note: IVF, *in-vivo* fertilization; DCDA, dichorionic diamniotic twins; NIPT, non-invasive prenatal testing.

Fetal gender uncertainty: refers to the loss of visits to foreign induced labor or failure to identify fetal sex after induced labor.

cal analysis showed significant differences in concomitant cardiac anomalies between the groups ($p < 0.001$; Tables 2,3,4).

4. Discussion

4.1 Fetal Situs Inversus

Complete situs inversus, also referred to as total situs inversus, is a condition characterized by a "mirror-image" configuration of the thoracic and abdominal organs relative to their normal anatomical positions. This condition is also known as mirror-image dextrocardia. If complete situs inversus is not associated with organ or system malformations, most of the fetuses affected have a good prognosis. However, if accompanied by complex cardiac malformations, the prognosis is poor, which is associated with significant surgical challenges and complications.

Following the prenatal ultrasound screening guidelines [7], the multiplanar continuous scanning method is employed during routine prenatal ultrasound examinations. Initially, the fetal orientation is determined, which is key to identifying the location of the fetal viscera. The fetal orientation was determined based on the relationship between the fetal head and spine, with clockwise rotation indicating the left to right side of the fetus in the cephalic presentation and counterclockwise rotation indicating the left to right side in the breech presentation. The standard transverse section of the fetal abdomen was used to observe the positions of the fetal stomach bubble, abdominal aorta, and inferior vena cava.

Fetal orientation determination methods: in the left occiput anterior position, the spine is on the left, and the stomach bubble is in the far field. In the right occiput anterior position, the spine is on the right, and the stomach bubble is in the near field. In the left sacrum anterior position, the spine is on the left, and the stomach bubble is in the near field. In the right sacrum anterior position, the spine is on the right, and the stomach bubble is in the far field. If the fetal position is suboptimal (oblique or transverse), maternal movement is encouraged, and re-examination is performed when the fetal position is suitable.

Between May 2018 and September 2023, our center correctly diagnosed 20 mid-trimester fetuses with situs inversus (no missed or misdiagnosed cases). This achievement can be attributed to several factors. First, 8 of the 20 cases (40.00%) were referred after initial external diagnosis, reflecting our center's role as a regional referral hub. Second, all patients underwent mandatory screening supervised by 2 senior ultrasonographers, both holding prenatal diagnostic certification and associate/full professor titles. Finally, real-time consensus verification was implemented for every case.

This study included 43 fetuses with visceral situs anomalies, among which 20 (46.51%) were diagnosed with complete situs inversus totalis. Notably, only 1 of these fetuses (5.00%) presented with complex cardiac anomalies, consistent with previous literature reports [8]. When fetal situs inversus is detected during prenatal ultrasound examination, a thorough evaluation, including ventricular looping patterns, atrioventricular connections, ventriculoarterial alignments, and spatial relationships of the great arteries, must be conducted to identify any associated intracardiac malformations. To assess potential extracardiac anomalies, a comprehensive fetal scan should also be performed.

4.2 Fetal Heterotaxy Syndrome

Fetal heterotaxy syndrome, also known as partial situs inversus, may involve only the abdominal or thoracic organs. When the viscera are in the normal position, but the heart is positioned in the right thorax, this condition is referred to as dextroversion. Conversely, if the viscera are arranged in a mirrored configuration, but the heart is located in the left thorax, this condition is known as levoversion. The International Nomenclature Committee for Pediatric and Congenital Heart Disease recommends classifying these conditions as visceral ectopia, which is also known as cardiosplenic syndrome [9,10].

Table 2. Comparison of cardiac malformations in fetal visceral anomalies.

Type	Total (n = 43)	Normal (n = 19, %)	Abnormal (n = 24, %)	*p*-value [a]
Situs inversus	20	19 (95.00%)	1 (5.00%)	<0.001
Heterotaxy syndrome	23	0 (0.00%)	23 (100.00%)	

[a]Fisher's Exact Test.

Table 3. Distribution of cardiac structural abnormalities in fetal visceral anomalies.

Cardiac structural abnormalities	Situs inversus (n = 20)	Heterotaxy syndrome (n = 23)
Pulmonary stenosis	1	7
Ventricular septal defect	1	4
Double outlet right ventricle	0	8
Complete transposition of the great arteries	1	5
Complete atrioventricular septal defect	0	4
Common arterial trunk	0	5
Interrupted inferior vena cava	0	3
Single atrium/single ventricle	0	13

Table 4. Distribution of cardiac structural abnormalities in cardiosplenic syndrome.

Cardiac structural abnormalitie	Left isomerism (n = 10)	Right isomerism (n = 13)
Interrupted inferior vena cava	3	0
Double superior vena cava	2	0
Anomalous pulmonary venous drainage	1	0
Complete atrio-ventricular septal defect	2	2
Double outlet right ventricle	4	4
Ventricular septal defect	3	2
Pulmonary artery stenosis	3	4
Single atrium/single ventricle	3	10
Common arterial trunk	1	4
Complete transposition of the great arteries	1	4
Persistent left superior vena cava	0	1

Cardiosplenic syndrome is mainly divided into 2 types [11]: The first type is polysplenia syndrome, which is characterized by having multiple spleens, bilateral left atrial appendages (left isomerism), bilobed lungs (morphologically left lungs), and interrupted inferior vena cava. It is often associated with atrioventricular septal defects, double-outlet right ventricle, and other complex cardiac malformations, sometimes accompanied by heart block [12]. The other type is asplenia syndrome, which is characterized by the absence of the spleen, bilateral right atrial appendages (right isomerism), trilobed lungs (morphologically right lungs), and associated intracardiac malformations, such as having a single atrium, single ventricle, double-outlet right ventricle, and atrioventricular septal defects. In partial situs inversus, extracardiac malformations often include intestinal malrotation and abnormalities in the position and number of spleens [13,14]. The prognosis of partial situs inversus depends on the type and severity of the accompanying intracardiac and extracardiac malformations.

This study identified 23 cases of fetal visceral heterotaxy, all presenting with cardiovascular malformations (100.00%), which corresponds with published reports on the co-occurrence rates of these conditions [15]. The most common cardiac anomaly observed was a single atrium/ventricle (56.50%, 13/23), followed by double-outlet right ventricle (34.78%, 8/23) and pulmonary stenosis (30.43%, 7/23). Analysis by isomerism type revealed left isomerism in 10 (43.48%) cases, including 3 with a single atrium/ventricle, whereas right isomerism occurred in 13 (56.52%) cases, including 10 with a single atrium/ventricle. Notably, both groups had the same number of cases with double-outlet right ventricle (n = 4 each). This result is basically consistent with the reported incidence rate of fetal visceral ectopia combined with cardiovascular malformations [16,17].

Fetal partial visceral ectopia often presents with atrial isomerism, which highlights the importance of determining the left and right atria. The morphology of the atrial appendages is the most reliable basis for distinguishing between the left and right atria. In the four-chamber and short-axis views of the great arteries, the left atrial appendage usually appears finger-like, with a narrow base and slender shape, whereas the right atrial appendage is cone-shaped, with a broad base and shorter, thicker shape. However,

given the limitations of ultrasound technology, prenatal diagnosis of some signs of cardiosplenic syndrome remains challenging. Therefore, in addition to mastering the normal fetal anatomy, ultrasound physicians should employ a systematic scanning protocol to not only observe structural abnormalities but also clarify the relationship between fetal presentation and visceral position in order to avoid missing rare cases.

4.3 Primary Ciliary Dyskinesia

PCD, also referred to as Kartagener syndrome [18,19], is associated with recurrent respiratory infections after birth and reduced fertility in men. In this study, 15 cases with negative chromosomal karyotype were subjected to WES and microarray analysis. The results revealed 1 case with a suspected pathogenic variant of PCD-related genes (normal delivery) and 4 with variants of uncertain significance in PCD-related genes (1 induced labor and 3 normal deliveries). A report [20] suggests that PCD is a monogenic disorder with a low incidence, but with a genetic predisposition. Therefore, to increase the detection rates of PCD or other genetic abnormalities, WES is recommended for pregnant women with fetal visceral situs and normal chromosomal results, enabling early detection, treatment, and intervention.

4.4 Holt–Oram Syndrome

HOS, also known as atriodigital dysplasia syndrome or heart–hand syndrome, is a rare autosomal dominant monogenic disorder [21]. Typical clinical manifestations include congenital cardiovascular malformations and varying severity of unilateral or bilateral upper limb dysplasia. Common cardiovascular malformations include atrial and ventricular septal defects, patent ductus arteriosus, and tricuspid atresia. In this study, WES identified HOS in 1 of the 15 cases, which involved right isomerism with a single atrium, common atrioventricular canal, single outflow tract, and anomalous pulmonary venous drainage. No upper limb structural abnormalities were observed, and the pregnancy was terminated.

In this study, fetal visceral position abnormalities accounted for 0.59‰ (43/72,499) of the cases, significantly higher than the previously reported rates [1]. This discrepancy may be attributed to two factors: First, the study period (2018–2023) coincided with the coronavirus disease 2019 (COVID-19) pandemic, during which increased rates of first-trimester severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infection were observed. A previous study has suggested the potential contribution of viral infections to abnormal fetal visceral development [22]. Second, as a regional fetal medicine referral center, our hospital's case selection may be biased toward more complex or abnormal cases.

4.5 Study Limitations

This study has the following limitations: (1) Challenges in prenatal follow-up management due to the referral-based nature of our center's cases resulted in substantial lost-to-follow-up instances. (2) The rarity of fetal visceral heterotaxy limits the availability of reference literature for more in-depth comparative analysis. (3) The current evaluation is restricted to prenatal ultrasound diagnostic value without comparative studies with fetal magnetic resonance imaging (MRI) or autopsy findings (the latter remaining the gold standard for diagnosis). Future research should focus on: establishing a multicenter collaborative network to improve follow-up systems; conducting multimodal diagnostic comparisons integrating ultrasound, MRI and autopsy; developing a comprehensive diagnostic framework incorporating genetic evaluation.

5. Conclusions

Fetal visceral position abnormalities, particularly partial situs inversus, is a rare and complex congenital condition with significantly high morbidity and mortality rates owing to its strong association with severe cardiac anomalies. Seidl-Mlczoch *et al*. [23] demonstrated that accurate prenatal diagnosis enables the identification of high-risk fetuses and facilitates timely multidisciplinary interventions, thereby reducing postnatal care challenges and mitigating family burdens. Prenatal ultrasonography serves as a reliable diagnostic modality for detecting fetal situs inversus; thus, it not only aids in making informed pregnancy management decisions but also plays a crucial role in the clinical detection of birth defects.

A previous study has shown a significantly high recurrence risk (up to 10.00%) in pregnant women with a history of fetal visceral situs abnormalities, particularly partial situs inversus [1]. This critical prognostic information should be thoroughly discussed with patients, and enhanced prenatal monitoring is strongly recommended.

Availability of Data and Materials

All data generated or analyzed during this study are included in this published article.

Author Contributions

Study conception and design: XG, ML, PC and HW; data collection: XG, ML, GW, YC; analysis and interpretation of results: XG, ML. All authors contributed to editorial changes in the manuscript. All authors read and approved the final manuscript. All authors have participated sufficiently in the work and agreed to be accountable for all aspects of the work.

Ethics Approval and Consent to Participate

This investigation was approved by the Medical Science Ethics Committee of the International Peace Maternity

and Child Health Hospital, Shanghai, China (No: GKLW-A-2025-055-01). All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or research committee and with the 1975 Declaration of Helsinki, as revised in 2013. All patients or their families/legal guardians gave their informed consent for inclusion before they participated in the study.

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Conflict of Interest

The authors declare no conflict of interest.

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